Genetic Counselling for Ataxia (1/2)

INFORMATION FROM PATIENTS AND FAMILIES
A NATIONAL ATAXIA NETWORK INITIATIVE
MOVEMENT DISORDERS SOCIETY OF INDIA

Genetic counselling is a process that helps individuals and families understand conditions that are caused by changes in genes. It provides information, guidance, and emotional support to people who have, or may be at risk of having, a genetic ataxia. It involves discussing what the condition means, how it is inherited, the chances of passing it on, different available testing options, understanding the outcome of these tests, and what options are available for management or family planning.
The counselling session is tailored to your needs and disease presentation and may not be the same for two families, even with the same suspected genetic disease.

Why is genetic counselling important in ataxia?

1. *Understanding the cause*: Many families want to know why ataxia occurred. A genetic diagnosis provides clarity and may end years of uncertainty.
2. *Inheritance pattern*: Knowing whether the ataxia is inherited dominantly, recessively, or maternally helps identify who else in the family might be at risk.
3. *Family planning*: Couples who carry a faulty gene can make informed choices about having children. Options such as prenatal testing or preimplantation genetic testing (testing embryos before pregnancy) may be discussed.
4. *Early detection and management*: In some ataxias, early diagnosis helps manage complications or start supportive treatments sooner.
5. *Emotional support*: Counselling offers space to discuss fears, feelings, and coping strategies.

Steps involved in the genetic testing

Genetic testing is usually performed by a genetic counsellor, clinical geneticist, or your neurologist with expertise in genetics. It is essential to know about your disease, investigation details, previous reports, including genetic testing, if any, and details regarding your family pedigree to understand the possible inheritance pattern.
Genetic counselling not only helps in understanding the test reports but also why the test is required and what the options for testing are. The genetic counselling can be divided into:

1. Pre-test genetic counselling
2. Post-test genetic counselling

Pre-test genetic counselling

Pre-test genetic counselling is done before someone undergoes genetic testing. The objective of this step is to understand and communicate:

1. Information regarding what genes are, how they can cause disease when abnormal, and how they can be inherited.
2. What is the likelihood of a genetic cause of ataxia in the proband or a consultand? (A proband is an affected person in a family who is the first to be identified and brings the family to medical attention whereas a consultand is an unaffected person who seeks genetic counselling on behalf of an affected family member).
3. How it might be inherited (dominant, recessive, X-linked, or mitochondrial).
4. The risk to other family members.
5. The indications, pros, and cons of undergoing genetic testing.

Genetic Counselling for Ataxia (2/2)

INFORMATION FROM PATIENTS AND FAMILIES
A NATIONAL ATAXIA NETWORK INITIATIVE
MOVEMENT DISORDERS SOCIETY OF INDIA

6. Regarding different types of genetic testing available for your scenario and their strengths and limitations.
7. What are the possible outcomes of genetic testing?
8. Available support and management options if a genetic cause is identified
9. What are the next steps if the genetic test is negative or inconclusive

At the end of pre-test genetic counselling, the patient and the family members are expected to be able to make an informed decision regarding whether to go for genetic testing or not, and if yes, which test among the options provided to choose. To undergo testing is solely the decision of the patient (legal guardian if the patient is a minor or not in the capacity to provide consent), and the counsellor can only help them in making that decision.

Post-test genetic counselling

Post-test genetic counselling is done once the report of the genetic testing is available. This is important irrespective of the type of outcome of the genetic report, i.e., whether it is positive, negative, or inconclusive. The following aspects will be discussed in this step.

1. Describe in detail what is the outcome of the genetic report.
2. If *positive*:
   a. What does that mean to the patient?
   b. What does the report mean to the family with regard to the risk of inheriting the disease?
   c. What is the way forward with respect to available treatment options, support group, or enrollment in the ongoing studies/trials, subject to the patient's choice?
   d. Options of family planning with respect to pregnancy in a couple planning for pregnancy – options of prenatal testing, preimplantation genetic diagnosis etc.
3. If the report is *negative* or *inconclusive*:
   a. What does that mean with respect to the genetic nature of the disease
   b. Options of further tests to clear this uncertainty
   c. What does that mean for the risk of inheritance in future pregnancies/family members?

At the end of post-test counselling, the patient/family is expected to be able to reasonably understand their report and to make an informed decision based on that on the management plan.

Key points

- Genetic counselling is not just about testing, but it is about understanding, choice, and support.
- Through pre-test counselling, you learn what to expect before testing.
- Through post-test counselling, you understand the results and how to use that information wisely.
- Together, these steps help you and your family to make informed, confident, and compassionate choices about your health and future, and access the right support at the right time.
- Even if no cure exists, knowing the cause helps manage symptoms better.
- Counselling is confidential and respects your values and choices.